Researchers identify opportunities to advance genomic medicine

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Genetic discoveries over the past 25 years have substantially advanced understanding of both rare and common diseases, furthering the development of treatment and prevention for ailments ranging from

inflammatory bowel diseases to diabetes, according to a study published in *Nature* in January.

The paper, titled "A brief history of human disease genetics," reviews breakthroughs in the association of specific genes with particular disorders, progress mostly driven by advances in technology and analytical approaches. The study also provides a framework for medical innovation to improve clinical care in the field.

"The future of medicine will increasingly focus on delivering care that is tailored to an individual's genetic makeup and patterns," says Judy H. Cho, MD, Dean of Translational Genetics at the Icahn School of Medicine at Mount Sinai, Director of The Charles Bronfman Institute for Personalized Medicine, and a co-author of the report. "Applying this knowledge will help us to enhance personalized health and medicine for patients at The Mount Sinai Hospital now and for years to come."

The study tracks advances in genomics over the past two decades through better technology, expanded access to vast and diverse data, and the development of other foundational resources and tools. The researchers also note the evolution of how diseases were discovered and identified.

Another major advancement is the increasing availability of large prospective population-based cohorts, known as biobanks. These biobanks often include tissue samples from individuals of many ethnic backgrounds and provide access to a wide range of demographic, clinical, and lifestyle data. The study finds that systematic approaches to data sharing, such as global collaborative networks, are critical in characterizing new disorders.

Today, genetic testing for individuals with symptoms and for at-risk relatives occurs routinely; it ranges from cancer screenings to

noninvasive prenatal tests. But challenges remain, including the absence of evidence-based guidelines to support health care recommendations, disparities in testing across society, and the lack of experience in genomics by some health care professionals.

The researchers say the biggest task in the coming decade will be to optimize and broadly implement strategies that use human genetics to enhance understanding of health and disease, and maximize the benefits of treatment. This will require joint efforts by the industry and academia to establish:

- comprehensive inventories of genotype-phenotype relationships across populations and environments;
- proactive measures to address entrenched disparities in scientific capacity and clinical opportunities that benefit individuals and societies across the world;
- a systematic assessment of variant and gene-level function across cell types, states, and exposures;
- improved strategies for turning basic knowledge from assessments into fully developed molecular, cellular, and physiological models of disease development; and
- application of these biological insights to drive new treatment and preventive options.

More information: Melina Claussnitzer et al, A brief history of human disease genetics, *Nature* (2020). DOI: 10.1038/s41586-019-1879-7

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